FGFR4 gene

fibroblast growth factor receptor 4

Normal Function

The *FGFR4* gene provides instructions for making a protein called fibroblast growth factor receptor 4. This protein is part of a family of fibroblast growth factor receptors that share similar structures and functions. These receptor proteins play a role in important processes such as cell division, regulating cell growth and maturation, formation of blood vessels, wound healing, and embryo development.

The FGFR4 protein interacts with specific growth factors to conduct signals from the environment outside the cell to the nucleus. The nucleus responds to these signals by switching on or off appropriate genes that help the cell adjust to changes in the environment. In response, the cell might divide, move, or mature to take on specialized functions. Although specific functions of *FGFR4* remain unclear, studies indicate that the gene is involved in muscle development and the maturation of bone cells in the skull. The *FGFR4* gene may also play a role in the development and maintenance of specialized cells (called foveal cones) in the light-sensitive layer (the retina) at the back of the eye.

Health Conditions Related to Genetic Changes

prostate cancer

cancers

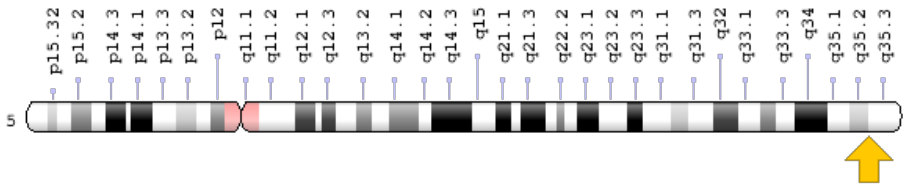
A variation (polymorphism) in the *FGFR4* gene that causes a switch in amino acids (the building blocks of proteins) is associated with several types of cancer, such as those that occur in the breast, colon, head and neck, and prostate. In people with this polymorphism, glycine is replaced by arginine at position 388 in the protein's chain of amino acids. This variation is common and appears to occur in about 50 percent of humans. Although it produces no ill effects in healthy people, the mutation is associated with accelerated disease progression in certain cancers.

The abnormal activation and increased activity of the *FGFR4* gene are also implicated in the development of pituitary tumors and gastric, pancreatic, and ovarian cancers.

Chromosomal Location

Cytogenetic Location: 5q35.2, which is the long (q) arm of chromosome 5 at position 35.2

Molecular Location: base pairs 177,086,872 to 177,098,144 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CD334
- FGR4_HUMAN
- hydroxyaryl-protein kinase
- JTK2 Gene
- protein-tyrosine kinase
- TKF Gene
- tyrosylprotein kinase

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FGFR4%5BTIAB%5D%29+OR+%28fibroblast+growth+factor+receptor+4%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- FIBROBLAST GROWTH FACTOR RECEPTOR 4
  http://omim.org/entry/134935

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/FGFR4ID512ch5q35.html
- HGNC Gene Family: CD molecules
  http://www.genenames.org/cgi-bin/genefamilies/set/471
- HGNC Gene Family: I-set domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/593
- HGNC Gene Family: Receptor Tyrosine Kinases
  http://www.genenames.org/cgi-bin/genefamilies/set/321
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3691
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2264
- UniProt
  http://www.uniprot.org/uniprot/P22455

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